

CASE REPORTS

CONGENITAL SKIN DEFECT OF THE NEWBORN

BY

O. D. BERESFORD, M.B., B.Ch., M.R.C.P.
Medical Registrar, Southmead Hospital, Bristol
and

P. D. SAMMAN, M.B., B.Ch., M.R.C.P.
Dermatological Registrar, University of Bristol

William Campbell (1826) published the first account of congenital skin defect of the newborn. The two patients he described were infants of the same family, the lesions affecting the scalp in each case; both infants died, one from haemorrhage on the eighteenth day, and the other from hydrocephalus at nine months; in the latter healing had begun before birth.

Jones (1849) described a patient with a defect extending from the third dorsal vertebra across the scapula and along the dorsal surface of the arm to within an inch of the elbow. Near the commencement was a nipple-like process of skin, and immediately beyond this healing had begun.

Since that time a number of cases have been reported in the literature, and Terruhn (1930) was able to collect seventy-nine in which the scalp was involved and twenty-nine involving other sites. Cases occurring subsequently have been reviewed by Freud et al. (1945).

The following three examples of this unusual skin defect have been seen in the past year, and it is considered that they are worth recording.

Case Histories

Case 1. R.C.M. was a full-term child born of healthy parents on March 7, 1947. Labour was normal, and there was no history of injury during pregnancy. There had been seven previous pregnancies: all of these children are now alive and well, and have no congenital malformations. The third child had been born two months prematurely and weighed only two and a half pounds at birth.

The midwife noticed an abnormality of the scalp and showed it to the mother a few minutes after birth: the child was not seen by us until three weeks later, and the mother stated there had been no material change during this time. There was a large skin defect on the vertex of the scalp behind the anterior fontanelle, extending more to the left than the right of the mid line, and roughly oval in outline, measuring seven by five cm., the larger measurement being antero-posterior; the edges of the lesion showed evidence of healing, and the remainder was covered by a scar. There appeared

to be a defect in the skull under this region, and radiologically this was shown to involve the left parietal bone and to a lesser extent the right. Anterior to the large lesion there were two smaller ones a few millimetres in diameter.

To the left of the skin defects a large dilated vein was visible below the normal skin and extending the whole length of the skull.

The blood Wassermann reactions of the mother and child were negative.

Subsequently healing progressed steadily. Portions of the scab fell off from time to time with consequent healing. By four months healing was complete with hair growing over much of the area.

Case 2. Baby S. was born six weeks prematurely on April 4, 1947. Labour was normal, and there was no history of injury during the pregnancy. There had been one pregnancy two years previously, and this child is alive and healthy with no congenital malformations. During pregnancy the mother had been healthy, and physical examination revealed no abnormality. Four years previously partial thyroidectomy had been performed because a large thyroid had caused respiratory embarrassment. The father also was healthy but he too had a partial thyroidectomy four years previously on account of thyrotoxicosis. An exhaustive family history failed to reveal any evidence of congenital malformations.

The appearance of the infant was striking. The skin was absent over the tip of the nose, the tips of both ears, and the adjacent area of the right occipital region. It was also absent over the dorsal surfaces of the hands and both surfaces of the wrists, and over the dorsum of the feet and anterior surface of the legs, knees, and lower thighs. The calves were covered with skin but the ankle regions were bare; skin was present over the palms of the hands and soles of the feet.

In the areas where the skin was lacking there was a membrane which was quite transparent at birth so that vessels, nerves, tendons, and muscles could be viewed clearly through it. During the course of the next few days this membrane was converted into a scab. No other congenital abnormalities were discovered, but the child's general condition was poor and it died on the fifth day in spite of

receiving all the care given to a premature infant. The blood Wassermann reactions of father, mother, and infant were negative. The specimen was preserved intact (figs. 1 and 2) and necropsy was not performed.

The infant measured, from crown to rump, 29 cm. The lesions measured:

Left arm and hand (dorsum) ..	6.3 cm.
Right arm and hand (dorsum) ..	6.3 cm.
Left thigh (anterior) to patella ..	5.7 cm.
Left patella to left external malleolus ..	8.2 cm.
Left external malleolus to extremity ..	5.0 cm.
(The right leg was similar)	
Right occipital region ..	2 by 1.5 cm.
Tip of nose ..	1.2 by 1.2 cm.
Lateral surface right elbow ..	0.6 by 0.6 cm.

HISTOLOGICAL EXAMINATION. A portion of skin was removed from the right forearm which included the transition from normal to bare area. Microscopically it showed complete absence of epidermis and epidermal structures over the affected area, the change from the normal being clearly marked; subcutaneous fat disappeared at the same point as the epidermis. The dermis remained intact, and as the subcutaneous fat was missing the dermis rested directly on the underlying muscle; blood vessels in the dermis were slightly dilated, and there was a mild infiltration with inflammatory cells, lymphocytes, and neutrophile polymorphonuclears (figs. 4 and 5).

Case 3. G.C. was not seen by us until he was two years old. He had an area of cicatricial alopecia on the vertex of the scalp slightly to the left of the mid line in the region of the lambda and measuring 5 by 2.5 cm.; there was no telangiectasia in connexion with it (fig. 3). The child also presented a much larger area of partial alopecia resulting from trichotillomania; mentally he was backward, but physically he appeared normal except for the skin lesion.

The mother said that at birth there had been a scabbed area on the scalp, much larger than the present scar, which had healed quite rapidly and much of it was soon covered with hairs. Birth had occurred at full term but it was a difficult breech delivery. The child weighed 7 lb. at birth, and throughout pregnancy the mother had been healthy and had not had any injury. There had been no other pregnancies and as far as could be ascertained there was no family history of other congenital malformations.

Blood Wassermann reactions of mother and child were negative.

Discussion

Congenital skin defect of the newborn is a rare abnormality affecting the scalp in the majority of cases; the lesions appear early in intra-uterine life and have been noted in a four-month foetus (von

Hoffmann, 1885). Ingalls (1933) described lesions seen in embryos as early as the second month and which he believed would have developed into the typical defect.

It is unusual for the abnormality to extend deeper than the epidermis, but on the scalp all structures down to the dura may be involved (Kehrer, 1910). The child may be stillborn or it may have other congenital malformations.

Clinically the condition at birth may show a raw granulating surface or it may be covered with a crust. Occasionally there may be a bulla or there may be a thin transparent membrane; healing may have started before birth. Later in life the condition shows as a scar.

Histologically there is a complete arrest of development of all layers of the epidermis and its associated glands and hair follicles (Emanuel, 1906). At the edges of the lesion the defect may be less severe, so that the final patch of alopecia is smaller than the raw area at birth.

The etiology of the condition is uncertain; an hereditary factor is present in a few cases (Campbell, 1826; Freud et al., 1945), but in the majority no such history can be obtained. There are two main views as to causation. The older maintains that it is due to amniotic adhesions; as a result of plastic inflammation of the amnion and epidermis, these grow together and cause arrest of growth of the epidermis. Later as the amniotic fluid increases, the two are forced apart and bands are formed; foetal movements assist in the separation, and the bands may separate entirely before birth. This theory is upheld by Kehrer (1910) and Terruhn (1930); the latter author describes one case in which such adhesions were present at birth, and shows with diagrams how the skin defect could be produced.

The second theory was advanced by Greig (1931) and amplified by Ingalls (1933). These writers believe the condition is due to an arrest of development; in favour of this is the usual mid-line position on the scalp, the striking symmetry in other areas, and the association with other congenital defects. Ingalls pointed out that the dorsal mid line, especially of the scalp, is subjected to important rearrangements during development so that abnormalities are most likely to be found here. He also points out that whilst the foetus is surrounded by fluid there is little tendency for the lesions to heal but that healing occurs rapidly after birth. A few cases may be the result of injury in utero (Kehrer, 1910) but many are not so caused, for mechanical injury would not be limited to the epidermis.

In reviewing the literature one is informed of the great similarity of cases. On the scalp they are

usually small and situated in the mid line of the vertex. On the trunk, lesions described by Hochstetter (1894) and Sutton (1935) are almost identical; whilst on the limbs the defects of the skin of the knees in the cases of Braun (1894) and Abt (1917) are similar. Our second case was similar to the one described by Rogatz and Davidson (1943).

No history of rubella or other infection was obtained in any of our cases, and it is our opinion that the condition is a congenital malformation possibly due to a failure of development of the superficial blood vessels over the affected area.

Summary

1. Three cases of a congenital ectodermal defect of the newborn are described.
2. The literature is reviewed, and theories of causation are outlined.

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FIG. 1.—Case 2.

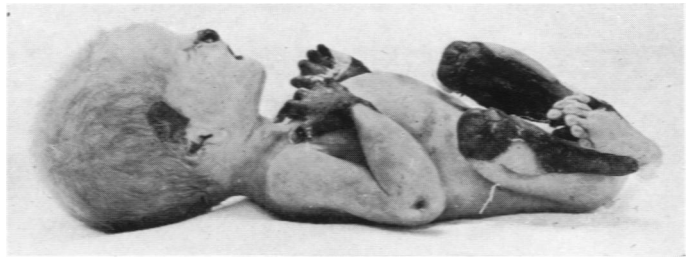
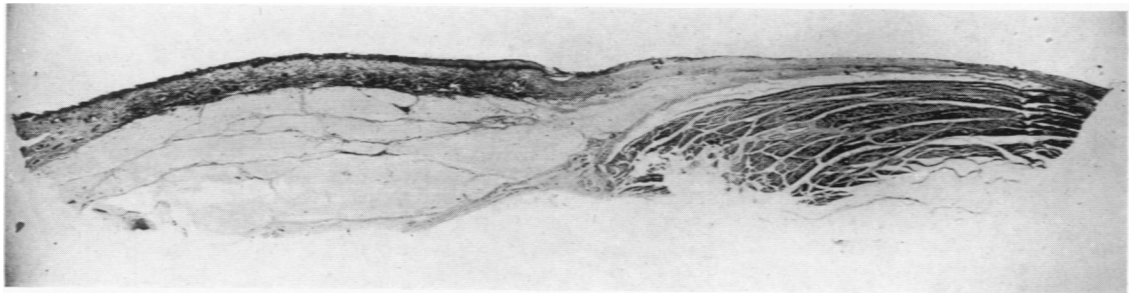


FIG. 2.—Case 2.



FIG. 3.—Case 3.

FIG. 4 (below.)—Case 2. Low power, showing loss of epidermal structure and subcutaneous fat.



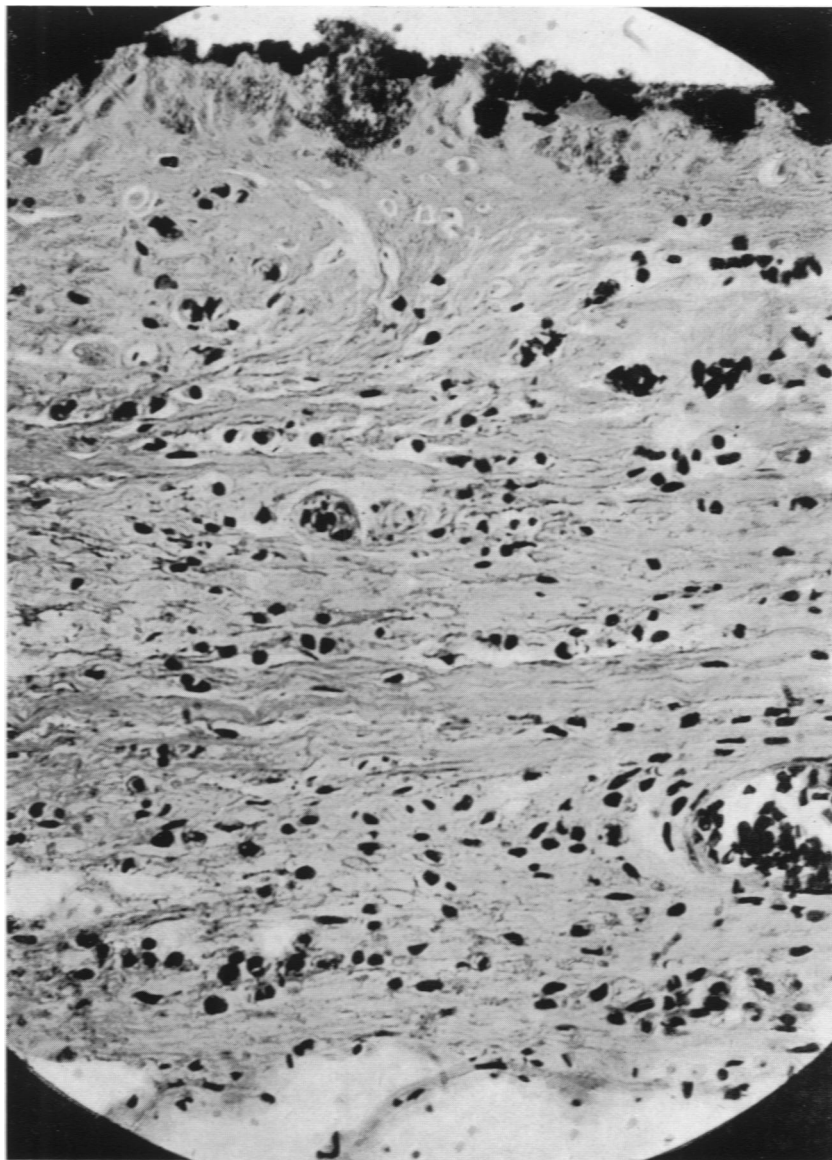


FIG. 5—Case 2. High power at point of transition showing last vestige of epidermis and infiltration of corium with leucocytes and lymphocytes.